- 1. Who was Gregor Mendel and what was his major contribution to science?
- 2. Draw Punnett Squares to show the outcomes of the following crosses:
  - A. AA X aa
  - B. Aa X aa
  - C. Aa X Aa

- 1. What is a test cross? How is it used?
- 2. R = tongue roller, r = nonrollerWhat would be the genotypic and phenotypic ratios for a cross between a heterozygous tongue roller and a non-roller?
- 3. Given: D = dimples, d = no dimples

What traits would the children of 2 parents (Rrdd and rrDd) have?

- 1. What is the probability that the following pair will produce the indicated offspring?
  - AABBCC X aabbcc -> AaBbCc
  - AABbCc X AaBbCc -> AAbbCC
- 2. Cross AaBb X Aabb. What is the probability of A\_B\_? That the baby will phenotypically resemble parent 1?
- Mom is A+. She has 2 children, one is O+ and the other is B-. (Note: Rh+ is RR or Rr, and Rh- is rr) What are the father's possible genotypes?

- 1. What is the probability that the following cross will produce the indicated offspring?
  - A. RR X Rr  $\rightarrow$  RR
  - B. AABB x aabb -> AaBb
  - C. AABbCc X AaBbCc -> AAbbCC
- 2. A couple has 2 children, both blonds with brown eyes. The parents are both brown eyed (BB), one with blond hair (rr) and one red (Rr). What is the probability the next child is a brown eyed redhead?

# Mendel And The Gene Idea CHAPTER 11



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## what you need to know:

- Terms associated with genetics problems: P, F<sub>1</sub>, F<sub>2</sub>, dominant, recessive, homozygous, heterozygous, phenotype, and genotype.
- How to derive the proper gametes when working a genetics problem.
- The difference between an allele and a gene.
- How to read a pedigree.
- How to use data sets to determine Mendelian patterns of inheritance.

Gregor Mendel

### Austrian monk

- Brought experimental and quantitative approach to genetics
- Bred pea plants to study inhertance
- Why peas?
  - Control mating (self- vs. crosspollination)
  - Many varieties available
  - Short generation time





#### EXPERIMENT



- P (parental) generation = true breeding plants
- F<sub>1</sub> (first filial) generation = offspring
- F<sub>2</sub> (second filial) generation = F<sub>1</sub> offspring

Table 11.1         The Results of Mendel's F1 Crosses           for Seven Characters in Pea Plants					
Character	Dominant Trait	×	Recessive Trait	F <sub>2</sub> Generation Dominant: Recessive	Ratio
Flower color	Purple	×	White	705:224	3.15:1
Seed color	Yellow	×	Green	6,022:2,001	3.01:1
Seed shape	Round	×	Wrinkled	5,474:1,850	2.96:1
Pod shape	Inflated	×	Constricted	882:299	2.95:1
Pod color	Green	×	Yellow	428:152	2.82:1
Flower position	Axial	×	Terminal	651:207	3.14:1
Stem length	Tall	×	Dwarf	787:277	2.84:1

## 7 characters in pea plants

# **Dominant** vs. **Recessive** (expressed) or (hidden)

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## Mendel's Principles

- 1. Alternate version of genes (alleles) cause variations in inherited characteristics among offspring.
- 2. For each character, every organism inherits one allele from each parent.
- 3. If 2 alleles are different, the **dominant** allele will be fully expressed; the **recessive** allele will have no noticeable effect on offspring's appearance.
- 4. <u>Law of Segregation</u>: the 2 alleles for each character separate during gamete formation.

## <u>Alleles</u>: alternate versions of a gene





**Dominant** (P), recessive (p)

- <u>homozygous</u> = 2 same alleles (PP or pp)
- heterozygous = 2 different alleles (Pp)



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- Phenotype: expressed physical traits
- <u>Genotype</u>: genetic make-up



## Punnett Square

• Device for predicting offspring from a cross

• Example: Pp x Pp (P=purple, p=white)

Genotypic Ratio:

Phenotypic Ratio:

<u>**Testcross</u>**: used to determine if *dominant trait* is unknown (homozygous or heterozygous?) by crossing with *recessive (pp)*</u>



### Law of Independent Assortment:

- Each pair of alleles segregates (separates) independently during gamete formation
- Eg. color is separate from shape



## Monohybrid Cross: study 1

character

• eg. flower color

<u>Dihybrid Cross</u>: study 2 characters

• eg. flower color & seed shape





# Dihybrid Cross

### • Example: AaBb x AaBb

## The laws of probability govern Mendelian inheritance

### • Multiplication Rule:

 Probability that 2+ independent events will occur together in a specific combination 
 multiply probabilities of each event

### • Ex. 1: probability of throwing 2 sixes

• 1/6 x 1/6 = 1/36

### • Ex. 2: probability of having 5 boys in a row

- $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{32}$
- Ex. 3: If cross AABbCc x AaBbCc, probability of offspring with AaBbcc is:
  - Answer: 1/2 x 1/2 x 1/4 = 1/16

## The laws of probability govern Mendelian inheritance

- Addition Rule:
  - Probability that 2+ mutually exclusive events will occur → add together individual probabilities
- Ex. 1: chances of throwing a die that will land on 4 or 5?
  - 1/6 + 1/6 = 1/3

# Segregation of alleles and fertilization as chance events



# Extending Mendelian Genetics

The relationship between genotype and phenotype is rarely simple

### Complete Dominance:

heterozygote and homozygote for dominant allele are indistinguishable

• Eg. YY or Yy = yellow seed

# **Incomplete Dominance**: F<sub>1</sub> hybrids have appearance that is between that of 2 parents

• Eg. red x white = pink flowers



**<u>Codominance</u>**: phenotype of both alleles is expressed

• Eg. red hair x white hairs = roan horses

### Multiple Alleles: gene has 2+ alleles

- Eg. human ABO blood groups
- Alleles =  $I^A$ ,  $I^B$ , i
- I<sup>A</sup>,I<sup>B</sup> = Codominant

(a) The three alleles for the ABO blood groups and their carbohydrates			
Allele	$I^A$	I <sup>B</sup>	i
Carbohydrate	A 🛆	В 🔾	none

#### (b) Blood group genotypes and phenotypes

Genotype	I <sup>A</sup> I <sup>A</sup> or I <sup>A</sup> i	I <sup>B</sup> I <sup>B</sup> or I <sup>B</sup> i	I <sup>A</sup> I <sup>B</sup>	ii
Red blood cell appearance				
Phenotype (blood group)	Α	В	AB	ο

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Blood Typing

Phenotype (Blood Group)	Genotype(s)
Туре А	I <sup>A</sup> I <sup>A</sup> or I <sup>A</sup> i
Туре В	I <sup>B</sup> I <sup>B</sup> or I <sup>B</sup> i
Type AB	[A]B
Type O	ii

## Practice Problem #1:

 A man who is heterozygous with type A blood marries a woman who is homozygous with type B blood. What possible blood types might their children have?

## Blood Transfusions



- Blood transfusions must match blood type
- Mixing of foreign blood  $\rightarrow$  clumping  $\rightarrow$  death
- Rh factor: protein found on RBC's (Rh+ = has protein, Rh- = no protein)





Africans (Zimbabwe)



SE Asians (Laos)



Native Americans



Indians



Australian Aborigines

# Practice Problem #2

- Babies Jane (blood type B), John (blood type O), and Joe (blood type AB) were mixed up in the hospital. Who are their parents?
  - Couple #1: A, A
  - Couple #2: A,B
  - Couple #3: B,O

**Polygonic Inhoritance:** the effect of 2 or more genes acting upon a single phenotypic character (eg. skin color, height)



# **Nature and Nurture:** both <u>genetic</u> and <u>environmental</u> factors influence phenotype



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Hydrangea flowers vary in shade and intensity of color depending on acidity and aluminum content of the soil.

## Mendelian Inheritance in Humans

<u>Pedigree</u>: diagram that shows the relationship between parents/offspring across 2+ generations



## Pedigree Analysis: Widow's Peak Trait



# Pedigree Analysis: PTC Tasting



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## Practice Problem #3

The pedigree below traces the inheritance of alkaptonuria, a biochemical disorder. Affected individuals are shaded. Does alkaptonuria appear to be caused by a dominant or recessive allele?



Genetic Disorders

### **Autosomal Recessive**

- Albinism
- Cystic fibrosis (CF)
- Tay-Sachs disease
- Sickle-cell disease
- Phenylketonuria (PKU)

### **Autosomal Dominant**

- Achondroplasia
- Huntington's disease (HD)
  - Lethal dominant allele

## Multifactorial Disorders

- Heart disease
- Diabetes
- Cancer
- Alcoholism
- Mental illnesses (schizophrenia, bipolar disorder)

# Genetic Counseling





Has genetic condition
No condition

## Practice Problems

 Cystic Fibrosis is an autosomal recessive disorder. What are the chances that 2 carriers for this disease will have a child with CF?

2. Huntington's Disease is an autosomal dominant disorder. If a woman with this disease marries a normal man, what are the chances that their children will have the disease?

Relationship among alleles of a single gene	Description	Example	
Complete dominance of one allele	Heterozygous phenotype same as that of homo- zygous dominant	PP Pp	
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	$ \begin{array}{c}                                     $	
Codominance	Both phenotypes expressed in heterozygotes	ΙΑΙΒ	
Multiple alleles	In the population, some genes have more than two alleles	ABO blood group alleles <i>I<sup>A</sup>, I<sup>B</sup>, i</i>	
Pleiotropy	One gene affects multiple phenotypic characters	Sickle-cell disease	

Relationship among two or more genes	Description	Example
Epistasis	The phenotypic expression of one gene affects the expression of another gene	$BbEe \times BbEe$ $BE bE Be be$ $BE bE Be be$ $BE bE A A A$ $BE A A A$ $A A A$ $BE A A A A$ $A A A A A$ $A A A A A$ $A A A A A A$ $A A A A A A A A A A A A A A A A A A A $
Polygenic inheritance	A single phenotypic character is affected by two or more genes	$\begin{array}{c c c c c c c c c c c c c c c c c c c $